Author Index to Volume 56

(ASHG) = American Society of Human Genetics report; (BR) = book review; (E) = editorial; (L) = letter to the editor; (PA) = presidential address

Abbas, N. E., 91
Abe, K., 374
Abeliovich, D., 1173
Adair, R., 692
Adès, L. C., 907
Ad Hoc Committee on Genetic Testing/Insurance Issues, 327 (ASHG)
Agid, Y., 193
Airò, P., 898
Aksentijevich, I., 1297
Albertsen, H., 484, 500
Ali, M., 1002 (L)
Allamand, V., 1417
Alley, T. L., 1411
Allotey, R., 336 (L)
Amalric, P., 396
Ambach, H., 854
Amyot, M., 1431
Anctil, J.-L., 1431
Anderson, L., 692
Andersson, B., 898
Andria, G., 1324
Antonarakis, S. E., 1502 (L)
Aoyama, T., 1287
Apold, J., 278
Arber, N., 1297
Arlett, C. F., 167
Arnqvist, H., 1075
Aro, A. R., 1493
Aronovich, E. L., 597
Arwert, F., 1096
Arzimanoglou, I., 544 (L)
ASHG Information and Education Committee, 535 (ASHG)
Ashizawa, T., 114, 1067
Athanasou, N., 1186
Atkinson, A. B., 1075
Auburger, G., 183
Audrézet, M. P., 272
Augarten, A., 1359
Aula, P., 1238 (L), 1493
Avner, E. D., 1101
Axelman, K., 332 (L)
Aymé, S., 542 (L)
Ayuso, C., 216
Babul, R., 243
Baciliero, U., 337 (L)
Bady, B., 374
Bailey, L. C., 938
Baker, E., 907
Baldini, A., 1502 (L)
Baldwin, C. T., 692
Ball, D., 336 (L)
Ballard, L., 484
Baranovskaya, S. S., 278
Barbier, N., 608
Bar-David, S., 1173
Bardoni, B., 862
Barkardottir, R. B., 254
Barrow, M. A., 400
Barth, P. G., 44, 663
Barton, D., 67
Basun, H., 332 (L)
Beckmann, J. S., 1417
Beiguelman, B., 1179
Belal, S., 1116
Bellus, G. A., 368
Benet, J., 452
Bengtsson, U., 1162
Ben Hamida, C., 1116
Ben Hamida, M., 1116
Bennett, C. P., 58
Bennett, R. L., 745
Benomar, A., 1116
Berg, M. A., 1287
Berger, R., 381
Bergoffen, J., 676
Bernal, J., 216
Bernasconi, F., 444
Bertario, L., 1060
Besser, G. M., 944
Betz, R., 1443
Bhattacharyya, S., 979
Biancalana, V., 224, 999 (L)
Bianchi, N. O., 1236 (L), 1247 (L)
Bignon, Y.-J., 254
Binkert, F., 444
Birch, J. M., 254
Birchfield, R. I., 963
Bird, T. D., 1007 (BR) (erratum, 1512)
Bisgaard, L., 1060
Bishop, T., 265
Blanco, R., 339 (L)
Blanton, S. H., 1125
Bleyl, S., 408
Blom, H. J., 142
Boerkoel, C. F., 887
Boers, G. H. J., 142
Boerwinkle, E., 1379
Boles, D. J., 716
Bolhuis P. A., 44, 1096
Bolognia, J. L., 58
Bonduelle, M., 272
Bonilla, E., 1017
Bonneau, D., 396, 542 (L)
Bonnet, D., 542 (L)
Borecki, I. B., 319, 1179
Borg, A., 254
Botta, E., 167
Bourge, U., 1417
Bowden, D. K., 294
Boyle, R., 1443
Brandi, M. L., 1075
Braun, A., 854
Braun, S. E., 597
Brautbar, C., 1359
Breast Cancer Linkage Consortium, 254, 265
Brenguier, L., 1417
Brenn, T., 1287
Breuning, M. H., 654, 663
Brice, A., 193, 1116
Brister, J. R., 75
Brody, L. C., 616
Broughton, B. C., 167
Broux, O., 1417
Brown, A., 630
Brown, E. M., 1075
Brown, R. H., 592
Brown, S., 1379
Brueton, L. A., 58, 938
Brugnoli, D., 898
Brzustowicz, L. M., 202, 210
Bu, X., 287
Buchanan, J. A., 243

Buetow, K. H., 310
Bunker, C. H., 461
Burghes, A. H. M., 158
Burke, W., 1007 (BR)
Burn, J., 58
Burt, M. J., 428
Buu, N. T., 845

Cahalane, S. F., 278
Cai, W., 1005 (L)
Callen, D. F., 663
Campbell, H. D., 175
Campeau, E., 243
Campiotto, S., 99
Camuzat, A., 396
Cancel, G., 193
Cann, H. M., 193
Cann, R. L., 349 (BR), 1248 (L)
Cappelli, M., 760
Cardon, L. R., 1224
Carey, J. C., 1249 (BR)
Carinci, F., 337 (L)
Carlson, C., 1391
Carpenter, N., 1304
Carr, A., 1186
Carreño, H., 339 (L)
Carrington, M., 1350
Carter, T. A., 202
Cartwright, P., 500
Casals, T., 623
Caskey, C. T., 114
Cats, A., 1060
Cavagni, G., 898
Cavalli-Sforza, L. L., 979
Ceballos-Picot, I., 592
Cellier, M., 845
Cerqueira, A., 99
Chakraborty, R., 461
Chalifaux, M., 1147
Chamberlain, S., 336 (L)
Chandrashekharappa, S., 705
Chang-Claude, J., 254
Chansky, H., 1132
Charmley, P., 963
Chen, K.-S., 175
Cherney, S. S., 1224
Chery, M., 1108
Chew, S. L., 944
Chiannilkulchai, N., 1417
Chiba, O., 1359
Chiffelle, I., 339 (L)
Childs, B., 359
Chillón, M., 623
Chneiweiss, H., 193
Chou, Y.-H. W., 1075
Chun, K., 558
Church, D. M., 1162
Cieply, K., 452
Cisternino, M., 862
Cleaver, J. E., 1257 (E)
Clegg, J. B., 294
Clerget-Darpoux, F., 1080
Cockburn, F., 278
Cohen, M. M., 1 (PA)
Cohen, D., 999 (L), 1417
Cohn, D. H., 388
Cole, D. E. C., 558, 880
Cole, J., 167
Collins, J., 1391
Concannon, P., 963
Conneally, P. M., 1417
Connolly, C., 216
Conrad, E. U., 1132
Cook, E. H., Jr., 993
Cooper, J. M., 1026
Cornelis, R. S., 254
Côté, G., 1431
Cote, G. J., 416
Cotton, R. G. H., 1034
Cox, D. W., 828 (E), 1140, 1315
Cox, N. J., 993
Cox, T. M., 1002 (L)
Cremers, C. W. R. J., 216
Croke, D. T., 278
Cruciani, F., 1005 (L)
Cullen, M., 1350
Cunniff, C., 676
Curioni, C., 337 (L)
Curran, J. L., 684
Curtis, D., 811 (L)

Dahl, H.-H. L., 553 (E)
Dahl, N., 224, 999 (L), 1108
Dai, X. Y., 335 (L)
Daly, M.J., 519
Damen, R., 183
Das, K., 210
Das, S., 570
Das Gupta, R., 1391
Davies, K., 67
de Andrade, M., 1379
DeCroo, S., 461
Deere, M., 698
Defesche, J. C., 1096
de Franchis, R., 1324
Deka, R., 461
Del Bono, E. A., 1240 (L)
Delhomme, B., 1367
Dell'Aquila, M., 676
Dembure, P. P., 630, 640
DeMeester, C. A., 287
Deng, G. Y., 528
Denning, C., 544 (L)
DePaulo, J. R., 1277
Der Kaloustian, V. M., 243
Deschamps, I., 1080
Deufel, T., 684, 1334
Devaud, C., 1417
Devilee, P., 254
de Vos, N., 654, 663
Devys, D., 106
Dharmavaram, R., 692
Dib, M., 592
Di Donato, S., 91, 1116
DiMauro, S., 1017
Djian, P., 1367
Doerflinger, N., 1116
Doggett, N., 663
Dooley, T. P., 663
Dörk, T., 623
Dorosz, S., 422
Doyle, J., 915
Dreyfus, M., 608
Dubourg, O., 193
Dubowitz, V., 151
Du Chesne, I., 1334
Duckett, D. P., 400
Dufier, J.-L., 396
Dunham, I. 1391
Dunn, M. A., 647
Durfy, S. J., 1008 (BR)
Dürr, A., 193, 1116
d'Urso, M., 1108

Easton, D. F., 254, 265
Echenne, B., 374
Eckhart, M., 1067
Ehrlich, P., 577
Eiken, H. G., 278
Eisensmith, R. C., 278
Elbaz, A., 374
Elkana, Y., 1194
Ellis, R., 684
Elsas, L. J. II, 630, 640
Elsner, T. I., 484, 500
Elston, R. C., 15 (E)
Engbersen, A. M. T., 142
Ergaz, Z., 1173
Erlich, H., 1350
Estivill, X., 623
Estop, A. M., 452
Evans, G. A., 676, 705
Evans, J. P., 342 (L)
Evinger, J., 630
Exelbert, R., 131, 887

Eyre, D. R., 388
Ezoe, K., 58

Falbella, R., 58
Fardeau, M., 374, 1108, 1417
Farrer, L. A., 692
Fasth, A., 898
Federico, A., 558
Feingold, E., 475
Feingold, N., 374
Feist, Y., 1334
Feitosa, M. F., 1179
Feldman, G. J., 938
Feng, Y., 106
Férec, C., 272
Ferlinz, K., 1343
Fernhoff, P., 630
Ferrell, R. E., 461, 812 (L)
Fex, J., 75
Figlewicz, D. A., 592
Fink, J. K., 188
Fisch, G. S., 1147
Fisher, J. M., 669
Flannery, A., 67
Flint, A., 926
Flint, J., 400
Florence, J., 158
Fodde, R., 1060
Fois, A., 558
Fonknechten, N., 623
Fontaine, B., 374
Fontaine, G., 616
Ford, D., 254, 265
Fougerousse, F., 1417
Francis, J. L., 428
Francke, U., 1287
Francomano, C. A., 368
Franken, D. G., 142
Frants, R. R., 99, 374
Frappier, D., 845
Fraumeni, J. Jr., 608
Frebourg, T., 608
Fridovich-Keil, J. L., 630, 640
Friedlander, Y., 1194
Friend, S. H., 608
Frosst, P., 1052
Fryer, A., 1304
Fryns, J.-P., 67, 676
Fujii, H., 1243 (L)
Fujiwara, M., 845
Fujiwara, Y., 1267
Fukai, K., 1320
Fulker, D. W., 1224
Furthmayr, H., 1287

Gadgil, M., 979
Gallion, H. H., 254
Ganau, A., 151
Ganczakowski, M., 294
Garber, J. E., 254, 608
Gardiner, R. M., 654
Gärtner, J., 854
Garver, B. L., 1246 (L)
Garver, K. L., 1246 (L)
Gasner, C., 1287
Gasser, D. L., 938
Gatto, E. M., 58
Gécz, J., 907
Gedeon, A., K., 907
Gelernter, J., 1262 (E)
Gerber, S., 396
Gerken, S. C., 484, 500
Germino, G. G., 1101
Gersh, M., 1404
Gieselmann, V., 51
Gigarel, N., 542 (L)
Gilbert, F., 544 (L)
Gilbert-Dussardier, B., 542 (L)
Gilgenkrantz, S., 1108
Giliani, S., 898
Gill, T. J. III, 1456
Gilliam, T. C., 202, 210
Giménez, J., 623
Gispert, S., 183
Gitschier, J., 570
Glinski, L. P., 732
Goldberg, R., 1391
Goldgar, D. E., 254
Goldman, A., 1373
Goldwurm, S., 428
Golomb, E., 1297
Goltsov, A. A., 278
Goodart, S. A., 1404
Gos, A., 1502 (L)
Goshen, R., 1359
Gotto, A. Jr., 1379
Goutières, F., 1116
Gouyon, J. B., 1108
Goyette, P., 1052
Grace, E., 676
Graham, J. M. Jr., 676
Gray, B. A., 1411
Gray, R. J., 287
Grazia Sacco, M., 898
Green, H., 1367
Greenberg, F., 175, 1156
Greenberg, J., 216
Grewal, P., 67
Griffioen, G., 1060
Grimm, T., 1334

Gros, P., 845
Gruber, H. E., 388
Grundfast, K. M., 75
Grunfeld, J. P., 374
Guay-Woodford, L. M., 1101
Gudmundsson, G., 1140
Guillot, A. P., 1101
Guimaraes, J., 374
Guldberg, P., 278
Gunaratne, P. H., 175
Gunter, K., 1042
Guo, S.-W., 1468
Güttler, F., 278
Gyapay, G., 1116

Haines, J. L., 1240 (L)
Haites, N., 254
Hakonen, A., 1493
Halliday, J. W., 428
Hallman, D. M., 416
Halloran Blanton, S., 698
Hamada, T., 231
Hamanishi, J., 745
Hamann, U., 254
Hammer, M. F., 951 (erratum, 1512)
Han, F. Y., 592
Hanauer, A., 224
Hanna, M. G., 1026
Hansen, M. F., 1125
Hao, H., 1017
Harcourt, S. A., 167
Harding, A. E., 1026
Harel, D., 1297
Harris, D. J., 1404
Hartung, E. J., 684, 1334
Harvey, J. F., 669
Hassold, T. J., 915
Hastings, V. A. 760
Hattori, M., 335 (L)
Hausmanowa-Petrusewicz, I., 210
Heath, H. III, 944
Heath, B., 944
Hebert, J. M., 979
Hecht, J. T., 368, 698, 1125
Hefferon, T. W., 368
Heilinger, D., 684
Heine, U., 1505 (L)
Heinisch, J., 131
Heinisch, U., 51
Helliwell, T. R., 725
Hemann, M., 302
Hendy, G. N., 880
Hentati, F., 1116
Héon, E., 1431

Herrin, J., 1101
Heyberger, S., 224
Heyer, E., 970, 1450
Heyetens, L., 684
Hietala, M., 1493
Hill, V. M., 944
Hill, W. G., 18
Hirono, A., 1243 (L)
Hirono, K., 1243 (L)
Hirst, M., 67
Hjelm, L N., 630
Ho, H.-N., 1456
Ho, L., 58
Hodge, S. E., 33
Hoeijmakers, J. H. L., 167
Hoffman, E. P., 350 (BR)
Hogue, D., 1125
Hoheisel, J. D., 175
Holden, J. J. A., 1147
Holik, P., 484
Holleman, R., 1101
Holmberg, E., 1304
Holmes, S. A., 58, 1320
Honeyman, K., 422
Hopkins, S. D., 1101
Hopper, J. L., 753
Horai, S., 951 (erratum, 1512)
Hors, J., 1080
Horton, V. K., 732
Horton, W. A., 368
Howard-Peebles, P. N., 1147
Hu, L. J., 1108
Huff, V., 84, 944
Huggins, R. M., 753
Hulsebos, T. J. M., 1096
Hultner, M. L., 1257 (E)
Hundrieser, J., 461
Hunter, A. G. W., 760, 1009 (BR)
Hurwitz, R., 1343

Ihara, T. 231
Ing, P. S., 216
Irons, D. A., 1101
Irven, C., 1186
Ishii, A., 1243 (L)
Ishii, N., 58
Itoh, T., 1267
Iwabuchi, K., 231

Jackson, C. E., 944, 1417
Jacobs, P. A., 669, 915
Jaffe, N., 84
Janicic, N., 880
Jara, L., 339 (L)
Järvelä, I. E., 654
Jaspers, N. G. J., 438
Jazwinska, E. C., 428
Jenkins, T., 586, 1373
Jensson, Ó., 1140
Jesudasan, R. A., 705
Jimenez, S., 692
Jin, K., 1456
Jin, L., 461
Jones, M. C., 676
Jones, O. W., 676
Jones, S. M., 188
Jongmans, W., 438
Jonsson, J. J., 597
Jorde, L. B., 11 (E), 347 (BR)
Jouet, M., 1304
Jurkat-Rott, K., 374
Juvonen, V., 1238 (L)

Kääriäinen, H., 907
Kafert, S., 51
Kaitila, I., 368
Kakulas, B. A., 422
Kalman, Y. M., 1359
Kaltwaßer, P., 862
Kammerer, S., 854
Kaneko, A., 294
Kaplan, J., 396
Kaplan, N. L., 18
Kark, J. D., 1194
Kasaboski, A., 760
Kastner, D. L., 1297
Katz, G., 1297
Kawada, Y., 335 (L)
Kayden, H., 1116
Keating, M. T., 1156
Keinänen, M., 907
Keith, C. G., 616
Kelsell, D. P., 254
Kemp, S., 44
Kenwrick, S., 1304
Kere, N., 1243 (L)
Kerem, B., 1359
Kerem, E., 1359
Ketterling, R. P., 343 (L)
Khan, P. M., 1060
Khoury, M. J., 835
Kieffer, J. E., 993
Kimberling, W. J., 216
King, M. D., 167
Kioschis, A. Gal. P., 1108
Klausnitzer, M., 684
Kleibeuker, J., 1060
Kleijer W. J., 44
Kleyn, P. W., 202, 210
Klitz, W., 1350
Klockgether, T., 183
Koenig, M., 1116
Kohlschütter, A., 1116
Kolodner, R., 1250 (BR)
Kolski, C., 224
Koskinen, T., 1088
Kousseff, B., 676
Kozak-Ribbens, G., 684
Kozman, H., 422
Kozyra, A., 769
Krahe, R., 1067
Krasowski, M. D., 993
Kraus, J. P., 1324
Kreuz, F., 183
Krieger, H., 1179
Krizus, A., 592
Kruglyak, L., 519, 1212
Kucherlapati, R., 1391
Kukolich, M., 342 (L)
Kunkel, L. M., 202
Kuzmin, A. I., 278

Laing, B. A., 422
Laing, N. G., 422
Lakkis, L., 106
Lalande, M., 926
Lalwani, A. K., 75
Lamb, N. E., 475
Lamers, W. H., 381
Lamminen, T., 1238 (L)
Lander, E. S., 519, 1212
Landrieu, P., 1116
Lange, B. M., 188
Lange, K., 1506 (L)
Langer, L. O., 698
Langley, S. D., 630, 640
Lannfelt, L., 332 (L)
Lapie, P., 374
Laporte, J., 999 (L)
Larsson, C., 1443
Lasseter, V. K., 1502 (L)
Lawrence, E., 484
Lee, S. C., 428
Lee, S.-H., 1411
Lehman-Horn, F., 684
Lehmann, A. R., 167
Lehmann-Horn, F., 374, 1334
Le Merrer, M., 542 (L)
Lennon, J. C., 640
Lenoir, G., 374
Leonard, J. C., 926
Leonard, J. V., 400
Le Paslier, D., 999 (L), 1116
Leppert, M. F., 188, 408, 944
Lerer, I., 1173

Lesicki, A., 188
Leventhal, B. L., 993
Leverton, K., 400
Levinson, B., 570
Levinson, D., 1359
Levy, J., 898
Li, F. P., 608
Li, W., 1287
Li, Z., 544 (L)
Liao, D., 343 (L)
Liberman, U., 1297
Lien, L., 202
Ligtenberg, M. J. L., 44, 1096
Lilius, L., 332 (L)
Lindblad, K., 1443
Lindblom, A., 254
Linder, C., 1116
Lindhout, D., 1060
Lindsay, E. A., 1502 (L)
Links, T. P., 374
Lissens, W., 272
Liu, J., 845
Livingston, R. J., 577
Loader, S., 769
Lochner-Doyle, D., 745
Loesch, D. Z., 753
Lohman, P. H. M., 438
Long, J. C., 799
Lorda-Sanchez, I., 444
Lorenzetti, D., 91
Loughlin, J., 1186
Luande, J., 1320
Ludwig, K., 310
Lunardi, J., 684
Lungarotti, M. S., 1096
Lunt, P. W., 1101
Lupski, J. R., 91, 175
Lusis, A. J., 287
Luzzatto, L., 294
Lynch, H. T., 254
Lyonnet, S., 542 (L)

Macchi, P., 898
Machado, M., 368
MacKay, N., 558
Maclaren, N. K., 528
Maddalena, A., 1147
Madgar, I., 1359
Magenis, R. E., 1042
Maha, G. C., 1505 (L)
Maher, E., 67
Majander, A., 1238 (L)
Malingré, H. E. M., 381
Mallette, L. E., 944
Mandel, J.-L., 44, 999 (L), 1108, 1116
Manfredi, G., 1017
Mantuano, E., 898
Marazita, M. L., 350 (BR)
Margaritte-Jeannin, P., 1080
Marie, S. K., 99
Markel, D. S., 745
Márquez, C., 452
Martínez, I., 248
Martini, A., 216
Martorell, M. R., 452
Marx, S., 1075
Mason, G. C., 1505 (L)
Massa, H. F., 926
Mastaglia, F. L., 422
Mathew, P. M., 416
Mathieu, M., 224
Matseoane, D., 210
Matsushita, M., 1132
Mattei, M.-G., 542 (L)
Mattina, T., 676
Mazur, L. A., 640
McCaskill, C., 1156
McEwen, J. E., 1477, 1487
McIntosh, I., 368
McKeown, C., 1304
McMahon, F. J., 1277
Meijers-Heijboer, A., 1060
Meininger, V., 592
Melis, M. A., 151
Mendell, J. R., 158
Menko, F. H., 1060, 1096
Mercier, B., 272
Meredith, C., 422
Merriwether, D. A., 812 (L)
Meschino, W., 243
Meyers, D. A., 1251 (BR), 1277
Michaud, J., 616
Migeon, B. R., 647
Miklos, G. L. G., 175
Milani, M., 216
Miller, D. C., 1287
Miller, F. W., 887
Mitchell, G. A., 616
Mitchison, H. M., 654
Miwa, S., 1243 (L)
Modiano, D., 1005 (L)
Mohr, J., 1060
Mole, S. E., 654, 663
Molecular Genetic Study Group, 760
Möller, C., 216
Monckton, D. G., 114
Moncla, A., 1304
Monnier, N., 374
Monsieurs, K., 684
Moore, M., 484
Moraes, C. T., 1017
Morali, F., 898
Moreno, F., 248
Morgan, K., 845
Morgan-Hughes, J. A., 1026
Morissette, J., 1431
Morris, G. E., 725
Morris, M. A., 1502 (L)
Morrow, B., 1391
Morton, N. E., 669
Moss, C., 58
Mountain, J. L., 979
Muecher, G., 1101
Mueller, C. R., 684
Muenke, M., 938
Mugneret, F., 1108
Muir, A., 528
Müller, C. R., 1334
Mulley, J. C., 907
Munné, S., 452
Munnich, A., 396, 542 (L)
Munsat, T. L., 202, 210
Muntoni, F., 151
Murray, J. C., 310

Nagengast, F., 1060
Naidu, S., 647
Nakajima, H., 131
Nanko, S., 335 (L)
Narod, S. A., 254
Natowicz, M. R., 870
Naughten, E., 278
Navarro, J., 452
Neel, J. V., 538 (L)
Nelson, D. L., 1147
Nelson, I., 1026
Nelson, L., 408
Nestadt, G., 1502 (L)
Nicastri, C., 887
Nichols, R., 887
Nickerson, E., 1156
Nicole, A., 592
Niebuhr, E., 1162
Nielsen, K. V., 1162
Niemelä, P., 1493
Nikali, K., 1088
Nikoskelainen, E., 1238 (L)
Nissim-Rafinia, M., 1359
Nivelon-Chevallier, A., 1108
Nivet, H., 374
Notarangelo, L. D., 898

Novelli, G., 272
Nunes, V., 623

O'Sullivan, C. K., 745
Obici, S., 202
Obie, C., 616
O'Connell, J. R., 1506 (L)
Odelberg, S. J., 408
O'Donnell, H., 1391
Ogunniyi, A. O., 1067
Oley, C. A., 400
Olkon, D. M., 993
Olson, J. M., 788
Omenn, G. S., 548 (BR)
Onari, K., 231
O'Neill, C., 278
Ono, T., 1267
Ophoff, R. A., 374
O'Rawe, A. M., 654
Orioli, I. M., 1207
Orozco, G., 183
Ortiz de Luna, R. I., 368
Oshimura, M., 438
Osuntokun, B. O., 1067
Otterud, B., 188, 408
Ottman, R., 821 (E)
Ottolenghi, C., 979
Ouahchi, K., 1116
Oude Luttikhuis, M. E. M., 400
Overhauser, J., 926, 1404

Packman, S., 570
Padberg, G. W., 99, 374
Padula, E., 337 (L)
Paglinauan, C., 1240 (L)
Palamino, H., 339 (L)
Palmer, S., 342 (L)
Pals, G., 1096
Pander, H.-J., 1245 (L)
Pandolfo, M., 91, 1116
Panico, M., 1324
Papapoulos, S. E., 1075
Paradis, A. J., 845
Parano, E., 202
Pareyson, D., 91
Passos-Bueno, M. R., 99
Pasturaud, P., 1417
Pasztor, L. M., 1404
Paterson, J., 1304
Patsch, W., 1379
Pauli, R. M., 698, 732, 1501 (L)
Pausova, Z., 880
Pavone, L., 202
Peltonen, L., 1088, 1493
Pena, S. D. J., 1503 (L)
Penchaszadeh, G. K., 202, 210
Penny, L. A., 676
Pepin, M., 1508 (BR)
Peral, B., 248
Pereira de Souza, A., 1417
Peterson, K. L., 1042
Petrova-Benedict, R., 558
Pezzetti, F., 337 (L)
Philip, N., 542 (L)
Pittelkow, M. R., 58
Piussan, C., 224, 374, 999 (L)
Piver, M. S., 254
Plaetke, R., 508
Plante, M., 1431
Plebani, A., 898
Ploplis, B., 75
Plotz, P. H., 131, 887
Pollak, M. R., 1075
Poll-The, B. T., 381
Polumbo, P. A., 388
Ponder, B. A. J., 254
Popovich, B. W., 1042
Porat, Y., 898
Powell, L. W., 428
Pramatarova, A., 592
Pras, E., 1297
Pras, M., 1297
Prence, E. M., 870
Primack, W., 1101
Prior, T. W., 158
Procaccio, V., 684
Proia, R. L., 716
Province, M. A., 319
Pulver, A. E., 1502 (L)
Pyper, W. R., 428

Quan, F., 1042
Quittell, L., 544 (L)

Raben, N., 131, 887, 1297
Rabl, W., 854
Rahat, A., 1359
Rahman, R. A., 705
Ramos, D., 623
Ramsay, M., 586, 1373
Ramus, S. J., 1034
Rao, D. C., 319, 1179
Raskind, W. H., 1132
Rave-Harel, N., 1359
Raymond, V., 1431
Read, A. P., 400
Reilly, P. R., 1010 (BR), 1477
Reinglass, T., 188
Reiser, C. A., 732
Resta, R. G., 745
Reynolds, A., 816 (BR)
Reznick, I., 898
Richard, I., 1417
Richard, M., 870
Richards, C. S., 114
Rigault, P., 1116
Rimoin, D. L., 388, 824 (E)
Roa, B. B., 91
Robbins, P., 422
Roberts, E. A., 1315
Robertson, E., 558, 938
Robin, N. H., 938
Robinson, B. H., 558
Robinson, W. P., 444
Roewer, N., 1334
Rogucka, E., 753
Rolinksi, B., 854
Ropers, H. H., 1096
Roscher, A. A., 854
Rosenblatt, D. S., 1052
Ross, B. M., 202
Rossi, A., 542 (L)
Rothhammer, F., 812 (L), 1236 (L), 1247 (L)
Rotter, J. I., 287
Roudaut, C., 1417
Rouleau, G. A., 243, 592
Rowley, P. T., 347 (BR), 769
Rozen, R., 1052
Rozet, J.-M., 396
Rübsam, B., 1334
Ruteshouser, E. C., 84
Ruttenberg, H. D., 408

Sabouraud, P., 1116
Sacco, M. G., 898
San Agustin, T. B., 75
Sánchez, F. O., 845
Sander, A., 310
Sandhoff, K., 1343
Sandkuijl, L. A., 654
San Millán, J. L., 248
Santavuori, P., 654
Santolamazza, P., 1005 (L)
Santos, N., 183
Sarde, C.-O., 44
Sasaki, H., 231
Saudubray, J. M., 374
Saunders, G. F., 84
Savontaus, M.-L., 1238 (L)
Scambler, P. 1391
Scapoli, L., 337 (L)
Schachtel, G. A., 508
Schäfer, K.-L., 1334
Schalling, M., 1443

Schapiro, J. M., 1297
Schappert, K., 616
Scherer, G., 862
Scherer, S. W., 342 (L), 1404
Scherpbier-Heddema, T., 310
Schinzel, A. A., 444
Schmeckpeper, B. J., 647
Schmelzle, R., 310
Schmitt-Ney, M., 862
Schneider, S. S., 926
Schoute, F., 1096
Schulz, J., 183
Schuman, J., 1240 (L)
Schurr, E., 845
Schwartz, E. I., 278
Schwinger, E., 1245 (L)
Scott, C. I., 698
Scozzari, R., 1005 (L)
Scriver, C. R., 278, 359, 814 (BR)
Sebastio, G., 1324
Seidman, C. E., 1075
Seidman, J. G., 1075
Selig, S., 202
Sellitto, D., 1005 (L)
Serville, F., 542 (L)
Sghirlanzoni, A., 91
Shaffer, L. G., 175, 1156
Sham, P. C., 811 (L)
Sharp, G. B., 188
She, J.-X., 528
Shen, J. J., 915
Sherman, J. B., 131
Sherman, S. L., 123, 475, 915
Shprintzen, R., 1391, 1502 (L)
Shriver, M. D., 461
Shugar, A. L., 915
Siciliano, M. J., 1067
Sidaner-Noisette, I., 1108
Siegel-Bartelt, J., 938
Silber, S. J., 272
Silburn, P., 1443
Silverman, G. A., 926
Simpson, S. G., 1277
Singh, R., 630
Sinnreich, R., 1194
Sirotkin, H., 1391
Skamene, E., 845
Slatter, R., 67
Smeitink, J. A. M., 381
Smith, L., 577
Smith, S. A., 254
Smyth, E., 254
Snow, K., 1147
Sobol, E., 1506 (L)
Sobol, H., 254
Sokol, R., 1116
Sommer, S. S., 343 (L)
Souied, E., 396
Soukup, S. W., 915
Speed, T. P., 1456
Spencer, H. G., 434
Sperandeo, M. P., 1324
Spiegel, R., 131
Spritz, R. A., 58, 1320
Spurr, N. K., 254
Srivatsan, E. S., 705
Stanbridge, E. J., 438
Stawski, S., 1240 (L)
Steel, G., 616
Steele, E., 630
Stefanini, M., 167
Stein, M. A., 993
Steinhaus, K. A., 745
Stephens, K., 342 (L), 577
Stevanin, G., 193
Stevens, E. M. B., 142
Stevens, G., 586
Stewart, A. D., 684
Stine, O. C., 1277
Stöckler, S., 854
Strina, D., 898
Strong, L. C., 84, 1125
Struewing, J. P., 254
Strunski, V., 224
Stuhlmiller, G. M., 1505 (L)
Sudbrak, R., 684, 1334
Sumegi, J., 216
Suomalainen, A., 1088
Surh, L. C., 760
Sutera, C. J, 769
Sutherland, G. R., 907
Suzuki, K., 1343
Suzuki, T., 231
Suzuki, Y., 231
Svensson, E., 278
Sweeney, M. G., 1026
Sybert, V. P., 577
Sykes, B., 1186
Syme, J., 676
Szabó, J., 944

Tada, J., 231
Takada, A., 231
Tamayo, M. L., 216
Taschner, P. E. M., 654, 663
Tashiro, K., 231
Teh, B. T., 1443
Templado, C., 452
Terwilliger, J. D., 777, 1088
Thakker, R. V., 944
Thandi, I., 1379
Thanh, L. T., 725
thi Man, N., 725
Thibodeau, S. N., 1147
Thiele, H., 862
Thomas, G., 647
Thomas, G. R., 1140, 1315
Thomas, P. M., 416
Thompson, A. D., 663
Thompson, A. F., 167
Thompson, E. M., 58, 938
Thompson, G. N., 616
Thomson, P. D., 1101
Thorsteinsson, L., 1140
Tiller, G. E., 388
Tint, G. S., 1404
Tischfield, J. A., 1417
Tognon, M., 337 (L)
Tonin, P., 254
Torroni, A., 123, 1234 (L)
Toss, G., 1075
Town, M., 294
Trask, B. J., 926
Treacy, E. P., 278, 359, 1034
Trembath, R. C., 400
Tremblay, M., 970
Triggs-Raine, B., 870
Trijbels, F. J. M., 142
Trope, G. E., 1431
Tsui, L.-C., 342 (L), 1404
Tsukerman, G., L., 278
Tuchman, A., 544 (L)
Twells, R., 336 (L)
Tyfield, L. A., 278
Tynan, K., 1287

Uhrich, S. B., 745
Underhill, P. A., 979
Urbanek, M., 799

Vainzof, M., 99
Vale-Santos, J., 374
Valle, D., 616
Valverde, K., 544 (L)
van Aarem, A., 216
van Amstel, H. K. P., 381
van Beukering, J., 586
van Beurden, E. A. C. M., 381
Van Broeckhoven, C., 684
van den Broek, M., 1060
van den Engh, G., 926
van den Berge, I. E. T., 381
van der Hoeven. J. H., 374
van der Klift, H., 1060
van Geel, B. M., 44

Vanier, M. T., 1343
Van Kirk, V., 452
van Leeuwen, C., 1060
van Leeuwen-Cornelisse, I., 1060
van Oost, B. A., 44, 1096
Varesco, L., 1060
Varvil, T., 188
Vasen, H., 1060
Verhaegh, G. W. C. T., 438
Verlingue, C., 272
Verloes, A., 542 (L)
Vermeulen, W., 167
Vezzoni, P., 898
Vieland, V. J., 33
Vila, A., 374
Villa, A., 898
Villaba, F., 84
Vincent, V., 745
Viribay, M., 248
Vitale, E., 202, 210
Voit, T., 183
Voullaire, L., 676
Vulliamy, T. J., 294
Vulpe, C., 570

Wadelius, C., 1304
Wagener, D. K., 835
Wagner, M., 1125
Wagstaff, J., 302
Wakisaka, A., 231
Waldo, F. B., 1101
Wallace, D. C., 1234 (L)
Wallace, M. R., 1404
Walshe, J. M., 1315
Wang, C. H., 202, 210
Ward, K., 408
Ward, K. A., 58
Warren, S. T., 106, 123
Wasel, N., 870
Wasmuth, J. J., 1162
Watkins, D., 243
Watkins, P. J., 1026
Weatherall, D. J., 294
Webb, S. I., 428
Weber, B. L., 254
Weber, C. A., 167
Weeks, D. E., 1506 (L)
Wei, S., 963
Weiler, M., 1343
Weir, B. S., 18
Weis, M. A., 388
Weiss, L., 1404
Weissenbach, J., 183, 193, 248, 310, 396, 542 (L), 684, 1088, 1116, 1431
Werder, E. A., 444
Weston, M. D., 216
White, R., 484, 500
Whitley, C. B., 597
Whitney, S., 570
Whittemore, A., 254
Wiggs, J. L., 1240 (L)
Wijker, M., 1096
Wijmenga, C., 99
Wijnen, J., 1060
Wijsman, E. M., 342 (L), 577
Wikström, M., 1238 (L)
Wilcox, E. R., 75
Will, K., 623
Williams, B. J., 915
Williams, C. A., 1404
Williams, R. C., 799
Williams, M. J. M., 434
Williamson, R., 336 (L)
Wilson, L. C., 400
Wilton, S. D., 422
Winnard, A. V., 158
Wintzen, A. R., 374
Wolstenholme, J., 400
Wolyniec, P. S., 1502 (L)
Wong, L.-J. C., 114
Woo, S. L. C., 278
Wright, P. G., 760
Wu, C.-t. B., 188

Xu, J., 202
Xu, W., 336 (L)

Yahyaoui, M., 1116
Yamaizumi, M., 1267
Yan, Y.-x., 608
Yoshiki, T., 231
Young, I. G., 175
Yu, L. M., 461

Zackai, E. H., 938
Zan-Bar, I., 898
Zar, H., 544 (L)
Zarbo, R. J., 944
Zatz, M., 99
Zdzienicka, M. Z., 438
Zegers, B. J. M., 898
Zelante, L., 676
Zenger-Hain, J., 676
Zerres, K., 1101
Zerylnik, C., 123
Zhao, X., 484
Ziadni, A., 1359
Zierz, S., 854
Zingg, M., 310
Zipursky, A., 915
Zlotogora, J., 51, 341 (L), 1173
Zlotogorski, A., 577
Zonana, J., 1042
Zori, R., 1404

Subject Index to Volume 56

(ASHG) = ASHG background paper or report; (E) = editorial; (L) = letter to the editor

A226V mutation, 616
ABC transporter, 44, 854
achaete-scute homologue 1 gene, 336 (L)
Achondroplasia, 368, 732, 824 (E), 1501 (L)
Acid maltase, 887
Adrenoleukodystrophy, 44
Adult onset, 1431
Affected relative pairs, 1212
African Americans, 1207
Africans, 1373
Albinism, 1173
Albright hereditary osteodystrophy, 400
Aldolase B gene, 1002 (L)
Algorithms, 519
Allelic association, 461, 1315
Allelic expression, 1186
Allelic heterogeneity, 265, 1417
Alternative splicing, 887
Alu
  Insertion, 951 (erratum, 1512)
  Sequence, 880
Alzheimer disease, 332 (L)
Amerindians, 812 (L), 1234 (L), 1236 (L)
Amino acid
  Metabolism, 616
  Transporter, 1297
Amyloid precursor protein, 332 (L)
Amyotrophic lateral sclerosis, 592
Anal atresia, 1042
Analysis, 452
Ancestral haplotype, 428
Anesthesia, 684
Anticipation, 99, 183, 341 (L)
Apolipoprotein
  (a), 287
  B, 287
  E, polymorphism, 1379
APP gene, 332 (L)
Arthritis, 692
Arylsulfatase A, 51
Ascending aortic aneurysm, 1287
Ascertainment, 15 (E)
  Bias, 33
Ashkenazi Jews, 1297
Asians, 1367
Aspartylglucosaminuria, 1493
Association, 335 (L), 993
  Analysis, 528
Ataxia, 1116
Ataxia-telangiectasia, 438
Atherosclerosis, 1379
ATPase, 1315
  Subunit 6, 1238 (L)
Attention-deficit hyperactivity disorder, 993
Attitudes, 1493
Australia/Australians, 428, 1034
Autoimmunity, 963
Autosomal dominant
  Cerebellar ataxia type 1, 193
  Polycystic kidney disease type 2, 248
Autosomal recessive
  Inheritance, 396
  Polycystic kidney disease, 1101
Axonal degeneration, 188

*Bam*HI RFLP, 339 (L)
Batten disease, 654, 663
β-Amyloid, 332 (L)
β-Hexosaminidase, 870
β-Oxidation, 44
Biochemical phenotype, 630
Bipolar affective disorder, 335 (L), 1262 (E), 1277
Bivariate flow karyotyping, 926
Blotchy mouse, 570
Books reviewed, author(s)/editor(s):
  Andrews, L. B., 814
  Bridge, P. J., 350
  Buelke-Sam, J., 548
  Cavalli-Sforza, L. L., 349
  Davies, K. E., 1250
  Emery, A. H., 1007, 1508
  Friedman, J. M., 1249
  Fullarton, J. E., 814
  Goldberg, S., 1010
  Hanis, C. L., 347
  Holtzman, N. A., 814
  Jeeves, M. A., 1009
  Kimmel, C. A., 548
  Krawczak, M., 816
  Lappé, M. A., 1008
  Lassonde, M., 1009
  Majerus, P. W., 347

Menko, F. H., 1007
Menozzi, P., 349
Motulsky, A. G., 814
Murphy, T. F., 1008
Nienhuis, A. W., 347
Ott, J., 1251
Piazza, A., 349
Polifka, J. E., 1249
Schmidtke, J., 816
Sing, C. F., 347
Stamatoyannopoulos, G., 347
Terwilliger, J., 1251
Varmus, H., 347
Warren, S. T., 1250
Books reviewed, title:
Assessing Genetic Risks: Implications for Health and Social Policy, 814
Calculation of Genetic Risks: Worked Examples in DNA Diagnostics, The, 350
Callosal Agents: A Natural Split Brain? 1009
Culture Clash: Law and Science in America, 1010
Developmental Toxicology, 2d ed., 548
Diagnostic Criteria for Neuromuscular Disorders, 1007
DNA Fingerprinting, 816
Genetics of Colorectal Cancer for Clinical Practice, 1007
Genetics of Cellular, Individual, Family, and Population Variability, 347
Genome Rearrangement and Stability, 1250
Handbook of Human Genetic Linkage, 1251
History and Geography of Human Genes, The, 349
Justice and the Human Genome Project, 1008
Molecular Basis of Blood Diseases, The, 2d ed., 347
Muscular Dystrophy: The Facts, 1506
Teratogenic Effects of Drugs: A Resource for Clinicians (TERIS), 1249
Boys, 753
Brachydactyly, 400
Brazilians, 99, 1179
BRCA1, 254, 265
Breakpoints, 452
Analysis, 500
Breast cancer, 254
Bullae without trauma, 1096

CA repeat, 335 (L)
Loci, 461
Ca^{2+}-sensing receptor, 1075
Gene, 880
CACNL1A3, 374
Calcium channel, 374
Cancer susceptibility genes, 608
Carotid artery, 1379
Carrier screening, 544 (L), 769
Cartilage, 1186
Cat cry, 1404
Caucasian, 374
CD40 ligand gene, 898
Cerebellar ataxia, 336 (L)
Cervical cancer, 705
Cervical cord compression, 1501 (L)
Cervical myelopathy, 732
Cervicomedullary-junction compression, 732, 824 (E), 1501 (L)
CFTR, 623, 1359
Microsatellite, 272
Mutation, 272
Chibchan, 812 (L)
Chilean Aymara, 812 (L)
Cholesterol, 1411
Chondrocalcinosis, 692
Chondrodysplasia, 388, 698
Chondrosarcoma, 1125, 1132
Chromosome(s), 452
Transfer, 438
Chromosome 1, 216
Chromosome 1p13, 396
Chromosome 1q21-q31, 944, 1240 (L)
Chromosome 1q23-q25, 1431
Chromosome 2q, 845
Chromosome 2q37, 400
Chromosome 3q13.1, 684
Chromosome 4, 408
Chromosome 4q, 248
Chromosome 5, 1162
Chromosome 5p deletion, 1404
Chromosome 6p, 938
Chromosome 6p21.1-p12, 1101
Chromosome 8, 1125, 1132
Chromosome 8q, 692, 1116
Chromosome 9, 243
Chromosome $9p^-$ syndrome, 302
Chromosome 11, 1125, 1132
Chromosome 11p, 416
Chromosome 11q deletion syndrome, 676
Chromosome 11q13 deletion, 705
Chromosome 14, 422
Chromosome 14q, 183, 193, 231
Chromosome 14q, 193
Chromosome 15q, 188
Chromosome 16, 663
Chromosome 17p, 484
Chromosome 17p11.2-p12, 91
Chromosome 18q deletion breakpoints, 926
Chromosome $18q^-$ syndrome, 926
Chromosome 18q21.2-q.22.2, 926
Chromosome 19, 698
Chromosome 19p13, 1443
Chromosome 19q12-13.2, 1334

Chromosome 22q11, 1391, 1502 (L)
Chromosome X, 553 (E), 647
  Adrenoleukodystrophy, 854
  Bullous dystrophy, 1096
  Deafness, 224
  Dilated cardiomyopathy, 151
  Dominant disease, 553 (E)
  Hydrocephalus, 1304
  Hyper IgM, 898
  Inactivation, 553 (E), 647, 1108
  Spastic paraplegia, 1304
Chromosome Xq13-q21, 224
Chromosome Xq28, 907, 1108
Chromosome Y, 951 (erratum, 1512)
Cleft lip/palate, 337 (L), 339 (L)
Cleft palate, 1391
Cleidocranial dysplasia, 938
Clinical medical genetic workload, 760
Clinical variability, 99
Clinical phenotypes, 854
CLN3, 654, 663
Cockayne syndrome, 167, 1267
COL2A1, 388, 1186
Collagen, type II, 388
Colorectal cancer, 1060
Common ancestor, 1034
Community genetics, 1493
Comparative mapping, 845
Complementation gene 1, 1267
Complementing alleles, 630
Computer algorithm, 500
Computer programs, 1506 (L)
Confidence regions, 1212
Congenital bilateral absence of vas deferens, 272
Congenital bilateral aplasia of vas deferens, 1359
Congenital defect, 1207
Congenital heart defects, 408
Congenital myopathy, 1026
Consanguinity, 519
Contiguous-gene–deletion syndrome, 175
Contiguous-gene syndrome, 542 (L), 1156
Copper
  Metabolism, 828 (E)
  Transport, 1140, 1315
Copper-transporting ATPase gene, 570
Coronary artery disease, 287
Coronary heart disease, 1194
Corrected homogeneity test, 1456
Craniocervical junction, 732
Craniosynostosis, 302
Cri-du-chat syndrome, 1162, 1404
Crime, 1487
Cryptic exon, 623
CTG repeats, 114
Curriculum, 535 (ASHG)
Cystathionine β-synthase deficiency, 1320
Cystic fibrosis, 544 (L), 623
  Phenotype, 272
Cystinuria, 1297

D-loop 6-bp deletion, 812 (L)
D2H, 1297
De novo mutation, 1067
Deafness, 216, 999 (L)
Decreased penetrance, 408
del(17)(p11.2), 175
Deletion(s), 302, 343 (L), 542 (L), 999, 1042
  Hotspots, 84
  Mapping, 1108, 1404
  Syndrome, 1162
  Terminal, 1404
Deletion/insertion mutation, 84
Demographic dynamics, 970
Demographic history, 979
Denaturing gradient-gel electrophoresis, 272, 1060
Developmental delay, 907
DFN3, 999 (L)
  Gene, 224
Diabetes mellitus, 1017, 1026
Digenic inheritance, 1417
Dihydropyridine receptor, 374
Dilated cardiomyopathy, 151
Dinucleotide-repeat polymorphism, 461
Discordant MZ twins, 647
Disease
  Gene mapping, 202
  Mapping, 18
  Risk factors, 835
  Susceptibility, 528
Disomic homozygosity, 915
Distal myopathy, 422
Distal 5p, 1162
Distal 11q deletions, 676
DMPK, 123
DNA
  Analysis, 1324
  Banking, 1477, 1487
  Data banking, 1477, 1487
  Deletion, 91
  Diagnosis, 760
  Laboratories, 1477
  Mismatch repair, 1060
  Repair, 167
    Defect(s), 1257 (E), 1267
  Synthesis, 438
Dominance, 577
Dopamine transporter, 993
Down syndrome, 475, 915
  Leukemia, 915
Duchenne muscular dystrophy, 158, 725

Dwarfism, 698
Dysmorphism, 1162
Dystrophin, 151, 158, 725

E-M algorithm, 799
Ectrodactyly, 342 (L)
Education, 535 (ASHG)
Egf-like domain, 1287
Elastin, 1156
Endocrine tumor gene HRPT2, 944
Endogamy, 979
Epidemiology, 294
  Methods, 835
Epidermal cytoskeleton function, 577
Epidermolysis bullosa, 577, 1096
ERCC2 gene, 167
Eugenics, 1245 (L), 1246 (L)
Europeans, 278, 654
Evolution, 1373, 1450
  Human, 1005 (L)
Exact tests, 508
Exon/intron boundaries, 75
Exon-PCR, 67
Exon skipping, 558, 716, 725
Exostoses, 1125
Expanding population, 1450
Extended pedigrees, 33
Eye disorder, 1431

Facioscapulohumeral muscular dystrophy, 99
Factor IX deletions, 343 (L)
Familial amyotrophic lateral sclerosis, 592
Familial benign hypocalcemia, 1075
Familial breast cancer, 265
Familial hypocalciuric hypercalcemia, 880
Familial hypocalciuric hypocalcemia, 1075
Familial hyperinsulinism, 416
Familial ovarian cancer, 265
Familial periodic cerebellar ataxia, 1443
Family history, 745
FGFR3, 368
Fibrillin biosynthesis/deposition, 1287
Fibrillin mutation, 1287
Fibrinogen, 1194
Fine mapping, 777
Fine structure mapping, 310
Finland, 1493
Finnish disease, 654
FISH, 705, 926, 1156, 1404, 1411
5,10-Methylenetetrahydrofolate reductase, 142
FLI FISH, 175
Flightless-1 gene (*fli*1), 175
FMR1, 106, 1042
  Deletion, 67
  Gene, 67
Folic acid, 1052
Foramen magnum, 824 (E)
Forensics, 1487
Founder contribution, 1450
Founder effect, 51, 231, 374, 970
Founder mutation, 1140
FPCA, 1443
Fragile site, 676
Fragile X
  Mutation, 1147
  Syndrome, 106, 1042
Frameshift hypothesis, 158
FRAXA mental retardation, 1042
FRAXE, 907
French Canadians, 870, 970, 1450
FSP1, 183
Fundus flavimaculatus, 396

Galactose-1-phosphate uridyltransferase, 630, 640
Galactosemia, 630, 640
Gametic disequilibrium, 799
Gel-retardation assay, 862
Gene dosage, 175
Gene location, 428
Gene mapping, 11 (E), 216, 231, 684, 788
Gene mutation, 1075
Gene survival, 1450
Gene therapy, 359
Gene-environment interaction, 821 (E)
Genetic counseling, 332 (L), 745, 760, 769, 1101
Genetic contribution, 970
Genetic deletion, 725
Genetic distance, 461
Genetic epidemiology, 1179, 1379
Genetic exclusion, 1240 (L)
Genetic heterogeneity, 698, 1334
Genetic isolate, 1417
Genetic linkage, 692, 698, 944, 1096
Genetic map(s)/mapping, 202, 210, 500, 508, 698, 1101, 1212
Genetic models, 1080
Genetic polymorphism, 1367
Genetic screening, 870
Genetic testing, 327 (ASHG), 1493
Genome, 1468
Genomic deletion, 887
Genomic imprinting, 434
Genotype/phenotype correlation(s), 374, 553 (E), 597, 1052, 1147
Germ-line deletions, 343 (L)
Girls, 753
Glaucoma, 1240 (L)
GLC1A locus, 1431
Glucose 6-phosphate dehydrogenase deficiency, 294, 1243 (L)

Glycogen storage disease, 381
Glycogenosis, 887
  Type VII, 131
G_{M2} gangliosidosis, 870
Gyrate atrophy, 616

Haploinsufficiency, 175
Haplotype(s), 278, 586, 654, 799, 1034, 1067, 1116, 1315, 1359, 1373
  Analysis, 210, 428, 1234 (L), 1236 (L), 1391
  Diversity, 951 (erratum, 1512)
  Relative risk, 428
Haplotype-based haplotype relative risk, 993
Haseman-Elston, 1224
Hemizygosity, 542 (L)
Hemochromatosis, 428
Hemoglobinopathies, 769
Hemophilia B deletions, 343 (L)
Hereditary ataxia, 1088
Hereditary bullous dystrophy, 1096
Hereditary diseases, 970
Hereditary fructose intolerance, 1002 (L)
Hereditary metabolic disease, 359
Hereditary multiple exostoses, 1125, 1132
Hereditary neuropathy, liability to pressure palsies, 91
Heterogeneity, 254, 341 (L), 1359
HEXA, 716
Hexosaminidase, 716
High-resolution genetic maps, 484
Hindu caste system, 979
Hispanics, 544 (L)
HLA, 528, 1350
  Heterodimer, 1080
hMSH2, 1060
HNPCC (hereditary nonpolyposis colorectal cancer), 1060
Homeodomain, 75
Homocysteine, 142, 1052
Homocysteinemia, 1052
Homocystinuria, 1052, 1324
Homozygosity, 1173
  Mapping, 416, 519, 1088
Homozygote, 577
Human acid sphingomyelinase, 1343
Human genome, 484
Human origins, 1234 (L), 1236 (L)
HUMF13A01 STR locus, 1005 (L)
Hunter syndrome, 597
Hyperhomocysteinemia, 142
Hyper IgM, 898
Hyperinsulinemic hypoglycemia, 416
Hyperparathyroidism, 944
Hypervariable regions, 979
Hypokalemic periodic paralysis, 374

Iceland, 1140
Identification testing, 1487
Identity by descent, 788, 1468
Iduronate-2-sulfatase, 597
IGF-2, 434
Imprinting, 1207, 1262 (E), 1277
In vitro contracture test, 684
Inborn errors, 616
Inbreeding, 1111 (L), 1503 (L), 1505 (L)
India, 979
Informed consent, 1477
Initiation, 158
Insertion mutation, 880
Insulin-dependent diabetes mellitus, 528, 1080
Insurance, 327 (ASHG)
Intermediate filament, 577
Interval mapping, 1224
Interval-mapping approach, 788
Involucrin, 1367
Ionizing radiation, 438
Isodisomy, 647
Isolated cases, 99
Israeli, 1194
Italians, 91, 1324

Jacobsen syndrome, 676
Japanese, 231
  Origin, 951 (erratum, 1512)
Jaw tumors, 944
Jomon, 951 (erratum, 1512)
Juvenile onset, 1431

Keratin, 577
Kibbutzim family study, 1194
KIT proto-oncogene, 58

L1, 1304
Lactic acidemia, 558
Lactic acidosis, 553 (E)
Late onset, 396
Leber hereditary optic neuroretinopathy, 1238 (L)
Leprosy, 1179
LGMD2A, 1417
Li-Fraumeni syndrome, 608
Likelihood
  Methods, 18, 777
  Models, 33
Limb malformation, 341 (L), 342 (L)
Limb-girdle muscular dystrophy, 1417
Linkage, 254, 310, 1125, 1224
  Analysis, 11 (E), 193, 231, 243, 248, 422, 938, 1240 (L), 1334, 1443
  Disequilibrium, 11 (E), 18, 123, 210, 777, 993, 1088, 1116, 1350, 1417
  Mapping, 408, 475, 654
Lip pits syndrome, 310

Lipoprotein (a), 287
Liver phosphorylase kinase
  α Subunit, 381
  Deficiency, 381
LMP2/LMP7 genes, 528
Locus heterogeneity, 342 (L)
Loss of heterozygosity, 1125, 1132
Lynch syndromes, 1060
Lysosomal enzyme processing, 1340
Lysosomal storage disease, 51

Machado-Joseph disease, 193, 231
Macrophage protein gene, 845
Macular dystrophy, 396
Major environmental effect, 1194
Major histocompatibility complex, 528, 1456
Malaria, 294
Male sterility, 272
Malignant hyperthermia, 1334
  Susceptibility, 684
Marfan syndrome, 1287
Marker association segregation χ^2, 1080
Markov chain, 1468
Martin-Bell phenotype, 67
MASA syndrome, 1304
Maternal age, 669
Maternal effect, 1080
Maternal transmission, 1262 (E), 1277
Maximum likelihood, 799
Medical genetics, 542 (L)
Meiotic breakpoint(s), 500
  Analysis, 508
Meiotic errors, 669
Meiotic recombination breakpoint, 484
Melanocyte, 58
Mendelian transmission, 319
Menkes disease, 570, 828 (E)
Mental impairment, 67
Mental retardation, 400, 1162
Metachromatic leukodystrophy, 51
Methylenetetrahydrofolate, 1052
  Reductase, 1052
Methyltetrahydrofolate, 1052
Microdeletion, 400, 663
  Syndrome, 1156
Microsatellite(s), 193, 428, 1111 (L), 1315, 1503 (L), 1505 (L)
  Deletions, 343 (L)
  DNA marker, 542 (L)
  Marker, 337 (L)
Microsatellite-based fine mapping, 310
Mismatch distributions, 979
Mitochondria, bbb (E), 1277
Mitochondrial contribution, 1450
Mitochondrial encephalomyopathy, 1026
Mitochondrial myopathy, 1017
Modifier locus, 434
Modifying genes, 265
Molecular cytogenetics, 1156
Molecular definition, 1391
Molecular genetics, 630
  Services, 760
Mondini-like dysplasia, 224
Monoamine oxidase A gene, 335 (L)
Monoclonal antibody, 725
Monosomy, 444
Mortality, 824 (E)
Mosaicism, 444
Mouse natural resistance–associated macrophage protein, 845
mRNA processing, 716
mtDNA, 812 (L), 1017, 1026, 1234 (L), 1236 (L), 1238 (L), 1247 (L), 1248 (L)
  Diversity, 979
  Markers, 1234 (L), 1236 (L)
Mucopolysaccharidosis, 597
Multilocus haplotyping, 1506 (L)
Multiple alleles, 1002 (L)
Multiple epiphyseal dysplasia, 698
Multiple-locus haplotypes, 799
Multiple sclerosis, 963
Multipoint inteval mapping, 1224
Multipoint linkage analysis, 519, 788
Muscle disorder, 684
Muscle-PFK deficiency, 131
Muscular dystrophy, 151, 725
Mutation(s), 114, 332 (L), 586, 640, 898, 1002 (L), 1034, 1320, 1343
  Analysis, 381, 558, 597, 630, 854, 1334
  Detection, 1140, 1315
Mutation-drift models, 461
Mutation-selection balance, 434
Myokymia, 1443
Myotonic dystrophy, 114, 123, 1067, 1373
Myotubular myopathy, 1108
MZ twinning, 647

Nail-patella syndrome, 243
National Society of Genetic Counselors, 745
Native Americans, 1234 (L), 1236 (L)
Nazi medicine, 1245 (L), 1246 (L)
Neonatal severe hyperparathyroidism, 880
Nesidioblastosis, 416
Neural cell adhesion, 1304
Neurological disease, 188
Neuromuscular disorder, 202, 210
Neuronal ceroid lipofuscinosis, 654, 663
Neurosurgical decompression, 824 (E)
Niemann-Pick disease, 1343
Nigerian, 1067

Nitric oxide synthase gene, 336 (L)
Nondisjunction, 444, 669
Nonequilibrium, 18
Nonpenetrance, 1359
Nonstatistical computation, 500
Nonsyndromic cleft lip and palate, 310, 337 (L)
North Americans, 1002 (L)
NRAMP 1 polymorphisms, 845
Nuclear families, 319
Nucleotide sequencing, 963

OAT genes, 616
Obesity, 1042
Occipital horn syndrome, 570
Ocular hypertension, 1431
Oculocutaneous albinism type 2, 586, 1320
Ordering loci, 508
Ornithine-δ-aminotransferase, 616
Osteoarthritis, 692, 1186
Osteochondromas, 1125
Ovarian cancer, 254
Overexpression, 438
OXPHOS deficiency, 1238 (L)

P gene, 586
  Mutations, 1320
p53, 608
Parathyroid carcinoma, 944
Parent-of-origin transmission, 1147
Parental imprinting, 1080
Parental origin, 669
Paternity testing, 1111 (L), 1503 (L), 1505 (L)
PAX3, 75, 1173
PCR, 926, 963, 1111 (L), 1324, 1503 (L), 1505 (L)
Pedigree(s), 745, 1334, 1468, 1506 (L)
  Nomenclature, 745
Penetrance, 341 (L)
Perilymphatic gusher, 224
Peroxisomal disorder, 854
Peroxisome, 44
PFK-M gene, 131
Phenotype parameters, 359
Phenotypic complementation, 438
Phenotypic features, 1162
Phenylalanine hydroxylase, 278, 1034
Phenylketonuria, 278, 1034
Phosphofructokinase, 131
Phosphorylase kinase, 381
Photosensitive disorders, 1257 (E), 1267
Physical mapping, 202, 926, 999 (L)
Physician education, 769
Piebaldism, 58
Pigment-dispersion syndrome, 1240 (L)
Pigmentation, 58
PKD2, 248
Point mutation, 887
Polish families, 210
Polycystic kidney disease, 1101
Polydactyly, 1207
Polymorphic marker loci, 777
Polymorphism, 799, 951 (erratum, 1512), 963
Population(s), 1315
  Expansions, 979
  Genetics, 123, 294, 461
  Growth, 538 (L)
Positional cloning, 1212, 1411
Prediction, 1379
Premature termination, 716
Prenatal diagnosis, 1101
Prenatal screening, 769
Presidential Address, 1
Primary open-angle glaucoma, 1431
Privacy, 1477, 1487
Progressive mixed deafness, 224
Promoter, 151
Pseudo-pseudohypoparathyroidism, 400
Pseudodeficiency, 870
Pseudogout, 692
Puberty, 753
Pulmonary venous return, 408
Pulsed-field gel electrophoresis, 287, 705
Pyridoxine, 616
Pyruvate dehydrogenase E1α
  Deficiency, 553 (E)
  Subunit, 558

Quality assurance, 745
Quantitative traits, 319
  Loci, 1224
Quebec, 1450
  Population founders, 970

R408W mutation, 278
Random sampling, 319
rBAT, 1297
Receptor, 58
Recessive alleles, 799
Recessive traits, 519
Recombination, 11 (E), 669, 1116, 1350
Recombination, 669
Recurrent mutations, 368
Recurrent spontaneous abortion, 1456
Reduced penetrance, 342 (L)
Regulation, 1477
Repeat expansion, 1450
Reproductive failure, 1456
Resolution, 1212
Retina, 616
Retinitis pigmentosa, 216
Rett syndrome, 647

Revertant fibers, 158
  Muscle, 725
RFLP, 278, 339 (L), 705
Risk assessment, 732
RNA-splicing mutation, 388
RNase protection, 106
Ryanodine receptor mutation, 1334
RY(i) hairpin loops, 343 (L)
RYR1 gene, 1334

Saguenay region, 1450
Schizophrenia, 1502 (L)
Segmental aneusomy, 676
Segregation
  Analysis, 33, 319, 1179, 1194
  Distortion, 1207
Selection, 51
Sequence analysis, 484
Sequencing, 597
Sequential sampling, 33
Sex ratio, 1207
Sex reversal, 862
Shared-allele test, 1456
Short tandem repeats, 278, 1034, 1111 (L), 1505 (L), 1503 (L)
Sib pairs, 788, 1224
  Linkage analysis, 287
Silver staining, 335 (L)
Single-strand conformational analysis, 1287
Site mutation, 623
Skeletal maturity, 753
SLC3A1, 1297
Smith-Lemli-Opitz syndrome, 1411
Smith-Magenis syndrome, 175
SOD 1, 592
Solomon Islands, 1243 (L)
Somatic instability, 114
Somatic mutation, 158, 725
South America, 339 (L)
South American populations, 1247 (L), 1248 (L)
Southern African, 586
Southwest Pacific, 294
Spanish, 248
  Cystic fibrosis mutation, 623
Spastic paraplegia, 183, 188
Speech delay, 1162
Sperm, 452
Spielmeyer-Sjögren disease, 654, 663
Spinal cerebellar ataxia
  Type 2, 336 (L)
  Type 3, 193
Spinal cord, 188, 732
Spinal muscular atrophy, 202, 210
Splicing, 388
  Mutation(s), 570, 887
Split hand/split foot, 341 (L), 342 (L)
Spondyloepiphyseal dysplasia, 388
SRY
  Mosaic, 862
  Mutations, 862
SSCP, 854
Stargardt disease, 396
Statistical methods/tests, 15 (E), 508
Steel factor receptor, 58
Sterilization, 1245 (L), 1246 (L)
Stochastic integral, 1468
STR polymorphisms, 1005 (L)
Submicroscopic deletion, 907
Sudden infant death, 824 (E)
Superoxide dismutase gene, 592
Surgical intervention, 1501 (L)
Swedish, 332 (L)

T-cell receptor Vβ gene, 963
Tanzania, 1320
TAP2, 1350
Tarui disease, 131
Tay-Sachs disease, 716, 870
Terminal deletions, 1404
Testing strategy, 799
Testis-determining gene, 862
TGFα, 339 (L)
Thiamine pyrophosphate, 558
Time analysis, 760
Tomaculous neuropathy, 91
Transcription, 151
  mRNA stability, 106
Transcription-related disorders, 1257 (E)
Transforming growth factor alpha, 339 (L)
Transient leukemia, 915
Translation, 106
Translocation, 302, 1411
Transmission disequilibrium test, 811 (L)
Transmitting male, 106
Transplantation, 359
Treatment, 359
Trichothiodystrophy, 167
Trigonocephaly, 676
Trinucleotide repeat(s), 1067, 1147
  Disorders, 123
  Length, 106
Trisomy, 444, 475
Trisomy 18, 669
Trisomy 21, 475, 915
tRNA glutamic acid, 1017
  Mutation, 1026
Tuberculosis susceptibility, 845
Tumor, 1125
Tumor-suppressor gene(s), 84, 705, 1132
Type 1 diabetes, 1080

Type II collagen, 1186
Type V collagen, 243
Tyrosinase-positive oculocutaneous albinism, 586
Tyrosinase-related protein, 302

Unexplained infertility, 1456
Uniparental disomy, 647
Usher syndrome II, 216
UV
  Light, 167
  Sensitivity, 1257 (E)
UV^s syndrome, 1267

Van der Woude syndrome, 310
Vanuatu, 294
Vascular disease, 142
Vasculogenesis, 408
Velo-cardio-facial syndrome, 1391, 1502 (L)
Vitamin E, 1116
VNTR, 278

Waardenburg syndrome, 75, 1173
Williams syndrome, 542 (L), 1156
Wilms tumor, 84, 434, 944
Wilson disease, 828 (E), 1140, 1315
WT1, 84

X chromosome
  Inactivation, 553, 1108
  Linkage, 381, 558, 999 (L), 1108
    Adrenoleukodystrophy, 854
    Bullous dystrophy, 1096
    Deafness, 224
    Dilated cardiomyopathy, 151
    Dominant disease, 553 (E)
    Hydrocephalus, 1304
    Hyper IgM, 898
    Spastic paraplegia, 1304
Xeroderma pigmentosum, 167, 1267
XY female, 862

Y chromosome, 951 (erratum, 1512)
YAC, 926, 1411
  Cloning, 845
Yeast, 640
  Expression, 131